Agency for Healthcare Research and Quality
Advancing Excellence in Health Care

NATIONAL QUALITY MEASURES CLEARINGHOUSE

General

Title

Sickle cell disease (SCD): percentage of children who, having initially tested positive for SCD through newborn screening, received confirmatory testing by 3 months of age.

Source(s)

Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC). Basic measure information: timeliness of confirmatory testing for sickle cell disease. Ann Arbor (MI): Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC); 2013 Oct 30. 40 p. [16 references]

Measure Domain

Primary Measure Domain

Population Health Quality Measures: Population Process

Secondary Measure Domain

Population Health Quality Measure: Population Access

Brief Abstract

Description

This measure is used to assess the percentage of children who, having initially tested positive for sickle cell disease (SCD) through newborn screening, received confirmatory testing by 3 months of age.

Rationale

In children with sickle cell disease (SCD), illness and death can be reduced through early diagnosis, treatment of complications, systematic follow-up and patient/parent education. In particular, timely confirmatory testing and prompt communication with families are steps that pave the way for initiating simple but life-saving treatments in young children with SCD. Neonatal screening for SCD is mandatory in the United States, but data are scarce that describe the proportion of children who receive confirmatory testing and the proportion of families who learn this information. This measure would highlight gaps where providers or health systems are falling short and encourage early and consistent treatment for all

young children with SCD.

Children with SCD benefit significantly from early diagnosis of disease. In a group of patients diagnosed with SCD as newborns, the mortality rate was 1.8% compared with an 8% mortality rate among those diagnosed after 3 months of age (Vichinsky et al., 1988). Most of the study was conducted prior to the use of prophylactic antibiotics, which today is a standard for pediatric SCD treatment and significantly reduces the frequency of life-threatening infections. The lower mortality rate in the study is attributed to early diagnosis and treatment of complications, practices that are supported, in large part, by extensive follow-up and patient and parent education. Newborn screening for SCD followed by confirmatory testing for positive screening results, parental education and comprehensive care reduces morbidity and mortality in infants and children with SCD (National Heart, Lung and Blood Institute, 2002).

Evidence for Rationale

National Heart, Lung and Blood Institute (NHLBI). The management of sickle cell disease. 4th ed. Bethesda (MD): National Institutes of Health, National Heart, Lung and Blood Institute, Division of Blood Diseases and Resources; 2002 Jun. 188 p.

Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC). Basic measure information: timeliness of confirmatory testing for sickle cell disease. Ann Arbor (MI): Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC); 2013 Oct 30. 40 p. [16 references]

Vichinsky E, Hurst D, Earles A, Kleman K, Lubin B. Newborn screening for sickle cell disease: effect on mortality. Pediatrics. 1988 Jun;81(6):749-55. PubMed

Primary Health Components

Sickle cell disease (SCD); screening; confirmatory testing; newborns; children

Denominator Description

The denominator is drawn from all sickle cell disease (SCD) cases reported in a state's newborn screening program records within the measurement year (see the related "Denominator Inclusions/Exclusions" field).

Numerator Description

The number of children who received confirmatory testing for sickle cell disease (SCD) by less than or equal to 90 days of age (see the related "Numerator Inclusions/Exclusions" field)

Evidence Supporting the Measure

Type of Evidence Supporting the Criterion of Quality for the Measure

A clinical practice guideline or other peer-reviewed synthesis of the clinical research evidence

A formal consensus procedure, involving experts in relevant clinical, methodological, public health and organizational sciences

A systematic review of the clinical research literature (e.g., Cochrane Review)

One or more research studies published in a National Library of Medicine (NLM) indexed, peer-reviewed journal

Additional Information Supporting Need for the Measure

Sickle Cell Disease Prevalence and Incidence
Sickle cell disease (SCD) is one of the most common genetic disorders in the United States (Kavanagh et al., 2011). The National Heart, Lung and Blood Institute (NHLBI) (2002) estimates that 2,000 infants are born with SCD in the United States (U.S.) each year. SCD affects 70,000 to 100,000 children and adults in the United States, predominantly those of African and Hispanic descent (Hassell, 2010).

Sickle Cell Disease Pathology and Severity
Vaso-occlusion (the sudden blockage of a blood vessel caused by the sickle shape of abnormal blood cells) is responsible for most complications of SCD, including pain episodes, sepsis, stroke, acute chest syndrome, priapism, leg ulcers, osteonecrosis and renal insufficiency (Steinberg, 1999). In addition, SCD can have hemolytic and infectious complications that result in morbidity and mortality in children with SCD (Kavanagh et al., 2011).

Sickle Cell Disease Burden in Daily Life
The effect of SCD on children and families is significant: severe pain episodes and hospitalizations restrict daily activities and reflect negatively on school attendance and performance, sleep, and social activities (Lemanek, Ranalli, & Lukens, 2009; Alvim et al., 2005). Although medical management of SCD continues to improve over time, 196 U.S. children died from SCD-related causes between 1999 and 2002 (Yanni et al., 2009).

Sickle Cell Disease Cost
In a study of health care utilization among low income children with SCD between 2004 and 2007, 27% of these children required inpatient hospitalization and 39% used emergency care during a year. Of these children, 63% averaged one well-child visit per year and 10% had at least one outpatient visit with a specialist (Raphael et al., 2009). Patients with SCD use many parts of the health care system, incurring significant costs. In 2009, mean hospital charges for children with SCD and a hospital stay were $23,000 for children with private insurance and $18,200 for children enrolled in Medicaid (HCUPnet, Healthcare Cost and Utilization Project, 2012). Kauf et al. (2009) estimate the lifetime cost of health care per patient with SCD to be approximately $460,000.

Performance Gap
There is significant variability and little tracking by more than half of states regarding this important issue. Neonatal screening for SCD is mandated in all 50 states, as well as in the District of Columbia, Puerto Rico, the U.S. Virgin Islands, and Guam. To our knowledge, there are no current published data that describe the proportion of children with a positive screen for SCD who receive confirmatory testing, or what proportion of families with a child who receives a positive confirmatory test result have results communicated to them. However, in a study of 52 State Newborn Screening Program follow-up coordinators with 100% participation, 100% of primary care providers were notified of positive screening SCD results, while only 81% of hematologists, 73% of hospitals, and 40% of families were notified of positive findings (Kavanagh et al., 2008). In programs where communication with the families was inconsistent, the responsibility for providing this information often fell upon someone other than the state newborn screening program. The efficacy of these efforts in ensuring timely confirmatory testing is unknown.

Evidence for Additional Information Supporting Need for the Measure

Alvim RC, Viana MB, Pires MA, Franklin HM, Paula MJ, Brito AC, Oliveira TF, Rezende PV. Inefficacy of piracetam in the prevention of painful crises in children and adolescents with sickle cell disease. Acta Haematol. 2005;113(4):228-33. PubMed

Hassell KL. Population estimates of sickle cell disease in the U.S. Am J Prev Med. 2010 Apr;38(4 Suppl):S512-21. PubMed

HCUPnet. Healthcare Cost and Utilization Project. [Web site]. Rockville (MD): Agency for Healthcare Research and Quality; 2006-2009

Kauf TL, Coates TD, Huazhi L, Mody-Patel N, Hartzema AG. The cost of health care for children and adults with sickle cell disease. Am J Hematol. 2009 Jun;84(6):323-7. PubMed

Kavanagh PL, Sprinz PG, Vinci SR, Bauchner H, Wang CJ. Management of children with sickle cell disease: a comprehensive review of the literature. Pediatrics. 2011 Dec;128(6):e1552-74.

Kavanagh PL, Wang CJ, Therrell BL, Sprinz PG, Bauchner H. Communication of positive newborn screening results for sickle cell disease and sickle cell trait: variation across states. Am J Med Genet C Semin Med Genet. 2008 Feb 15;148C(1):15-22. PubMed

Lemanek KL, Ranalli M, Lukens C. A randomized controlled trial of massage therapy in children with sickle cell disease. J Pediatr Psychol. 2009 Nov-Dec;34(10):1091-6.

National Heart, Lung and Blood Institute (NHLBI). The management of sickle cell disease. 4th ed. Bethesda (MD): National Institutes of Health, National Heart, Lung and Blood Institute, Division of Blood Diseases and Resources; 2002 Jun. 188 p.

Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC). Basic measure information: timeliness of confirmatory testing for sickle cell disease. Ann Arbor (MI): Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC); 2013 Oct 30. 40 p. [16 references]

Raphael JL, Dietrich CL, Whitmire D, Mahoney DH, Mueller BU, Giardino AP. Healthcare utilization and expenditures for low income children with sickle cell disease. Pediatr Blood Cancer. 2009 Feb;52(2):263-7. PubMed

Steinberg MH. Management of sickle cell disease. N Engl J Med. 1999 Apr 1;340(13):1021-30. PubMed

Yanni E, Grosse SD, Yang Q, Olney RS. Trends in pediatric sickle cell disease-related mortality in the United States, 1983-2002. J Pediatr. 2009 Apr;154(4):541-5. PubMed

Extent of Measure Testing

Reliability

Data/Sample. This measure is based on the gold standard data source for sickle cell disease (SCD): initial and confirmatory diagnosis information that is maintained by all state newborn screening programs in the United States. The Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC) tested this measure using newborn screening results from three state public health agencies: Illinois, Michigan, and Wisconsin. Newborn screening data captures the vast majority of births in states; in Michigan, 97% of all children have newborn screening performed, while in Illinois the figure is nearly 99%. The remaining births opt out of newborn screening based on religious grounds.

The measure was tested as specified, which requires an assessment among the entire population of a state's birth cohort that had an initial newborn screen indicating SCD. This measure includes no sampling; consequently, no sampling error is introduced that would necessitate the calculation of measure reliability.

Validity

The validity of this measure was determined through face validity established by a national panel of experts and advocates for families of children with SCD. Face validity is the degree to which the measure construct characterizes the concept being assessed, which was established by two Q-METRIC SCD expert panels. The panel established a very high degree of face validity for this measure through a detailed review of concepts and metrics considered to be essential to effective SCD management and treatment. The Q-METRIC expert panel included nationally recognized experts in SCD, representing hematology, pediatrics, and SCD family advocacy. In addition, measure validity was considered by experts in state Medicaid program operations, health plan quality measurement, health informatics, and health care quality measurement. In total, the Q-METRIC expert panels included 14 experts providing a comprehensive perspective on SCD management and the measurement of quality metrics for states and health plans.

From this group, concepts and draft measures were rated for their relative importance. This measure was among the most highly rated, with the expert panel's average score of 8.7 (out of a maximum of 9). Two rating methods were used to minimize any potential bias due to outlier ratings; this measure received identically high ratings (8.7) using both methods. In addition, the expert panelists noted that this measure not only was important, but could be accessed through state health departments. The measure as specified from state newborn screening program data was deemed to be the most valid, in contrast to candidate metrics that would be derived from provider data. Finally, the Q-METRIC SCD expert panel noted that this measure was a valid marker of important variations that they felt may exist across states; the panel noted that newborn screening programs can be highly variable across states and this measure would likely reflect those variations.

Evidence for Extent of Measure Testing

Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC). Basic measure information: timeliness of confirmatory testing for sickle cell disease. Ann Arbor (MI): Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC); 2013 Oct 30. 40 p. [16 references]

State of Use of the Measure

State of Use

Current routine use

Current Use

not defined yet

Application of the Measure in its Current Use

Measurement Setting

Ambulatory/Office-based Care

Community Health Care

Hospital Inpatient

Hospital Outpatient

Regional, County, or City Public Health Programs

State/Provincial Public Health Programs

Transition

Type of Care Coordination

Coordination between providers and community

Coordination between providers and patient/caregiver

Professionals Involved in Delivery of Health Services

not defined yet

Least Aggregated Level of Services Delivery Addressed

State/Provincial

Statement of Acceptable Minimum Sample Size

Does not apply to this measure

Target Population Age

Newborn

Target Population Gender

Either male or female

National Framework for Public Health Quality

Public Health Aims for Quality

Population-centered

National Strategy for Quality Improvement in Health Care

National Quality Strategy Aim

Healthy People/Healthy Communities

National Quality Strategy Priority

Effective Communication and Care Coordination
Health and Well-being of Communities
Prevention and Treatment of Leading Causes of Mortality

Institute of Medicine (IOM) National Health Care Quality Report Categories

IOM Care Need

Living with Illness

Staying Healthy

IOM Domain

Effectiveness

Timeliness

Data Collection for the Measure

Case Finding Period

The measurement year

Denominator Sampling Frame

Geographically defined

Denominator (Index) Event or Characteristic

Clinical Condition

Diagnostic Evaluation

Geographic Location

Patient/Individual (Consumer) Characteristic

Denominator Time Window

not defined yet

Denominator Inclusions/Exclusions

Inclusions
The denominator is drawn from all sickle cell disease (SCD) cases reported in a state's newborn screening

program records within the measurement year.

Note: Refer to the original measure documentation for codes to identify SCD.

Exclusions

Children who died within 120 days of birth are excluded from the denominator.
Children with diagnosis in the state newborn screening data indicating one of the SCD variants listed in Table 2 of the original measure documentation are specifically excluded from the denominator.

Exclusions/Exceptions

not defined yet

Numerator Inclusions/Exclusions

Inclusions
The number of children who received confirmatory testing for sickle cell disease (SCD) by less than or equal to 90 days of age

Note:

For the purposes of this measure, SCD is restricted to hemoglobin screening results for a subset of conditions considered to be clinically significant (refer to Table 1 in the original measure documentation).
Intake Period: January 1 of the measurement year to April 1 of the year following the measurement year.
Confirmatory Testing: Repeat testing performed to confirm initial newborn screening results. Confirmatory testing may be performed at any time following birth, using any of the methods shown in Table 1-A of the original measure documentation.

Exclusions
Unspecified

Numerator Search Strategy

Fixed time period or point in time

Data Source

State/Province public health data

Type of Health State

Does not apply to this measure

Instruments Used and/or Associated with the Measure

Unspecified

Computation of the Measure

Measure Specifies Disaggregation

Does not apply to this measure

Scoring

Rate/Proportion

Interpretation of Score

Desired value is a higher score

Allowance for Patient or Population Factors

not defined yet

Standard of Comparison

not defined yet

Identifying Information

Original Title

Timeliness of confirmatory testing for sickle cell disease.

Measure Collection Name

Sickle Cell Disease Measures

Submitter

Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC) - Academic Affiliated Research Institute

Developer

Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC) - Academic Affiliated Research Institute

Funding Source(s)

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Composition of the Group that Developed the Measure

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Financial Disclosures/Other Potential Conflicts of Interest

Unspecified

Adaptation

This measure was not adapted from another source.

Date of Most Current Version in NQMC

2013 Oct

Measure Maintenance

Unspecified

Date of Next Anticipated Revision

Unspecified

Measure Status

This is the current release of the measure.

The measure developer reaffirmed the currency of this measure in January 2016.

Measure Availability

Source available from the Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC) Web site . Support documents are also available.

For more information, contact Q-METRIC at 300 North Ingalls Street, Room 6C08, SPC 5456, Ann Arbor, MI 48109-5456; Phone: 734-232-0657; Fax: 734-764-2599.

NQMC Status

This NQMC summary was completed by ECRI Institute on July 25, 2014. The information was verified by the measure developer on September 16, 2014.

The information was reaffirmed by the measure developer on January 7, 2016.

Copyright Statement

Production

Source(s)

Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC). Basic measure information: timeliness of confirmatory testing for sickle cell disease. Ann Arbor (MI): Quality Measurement, Evaluation, Testing, Review, and Implementation Consortium (Q-METRIC); 2013 Oct 30. 40 p. [16 references]

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Readers with questions regarding measure content are directed to contact the measure developer.